CLAIMS

1. A method for detecting the presence or absence of a single nucleotide polymorphism (SNP) allele in a genomic sample, the method comprising:

preparing a reduced complexity genome (RCG) from the genomic sample, and

analyzing the RCG for the presence or absence of a SNP allele.

2. The method of claim 1, wherein the analysis comprises hybridizing a SNP-ASO and the RCG, wherein the SNP-ASO is complementary to one allele of a SNP, whereby the allele of the SNP is present in the genomic sample if the SNP-ASO hybridizes with the RCG, and wherein the presence or absence of the SNP is used to characterize the genomic sample.

3. The method of claim 2, wherein the RCG is immobilized on a surface.

4. The method of claim 2, wherein the SNP-ASO is immobilized on a surface.

5. The method of claim 2, wherein the SNP-ASO is individually hybridized with a plurality of RCGs.

6. The method of claim 1, wherein the RCG is a PCR-derived RCG.

7. The method of claim 1, wherein the RCG is a native RCG.

8. The method of any one of claims 1-7, wherein the method further comprises identifying a genotype of the genomic sample, whereby the genotype is identified by the presence or absence of the alleles of the SNP in the RCG.

9. The method of any one of claims 1-7, wherein the genomic sample is obtained from a tumor.

10. The method of claim 9, wherein a plurality of RCGs are prepared from genomic samples isolated from a plurality of subjects and the plurality of RCGs are analyzed for the presence of the SNP.

11. The method of claim 8, wherein the presence or absence of the SNP allele is analyzed in a plurality of genomic samples selected randomly from a population, the method further comprising determining the allelic frequency of the SNP allele in the population by comparing the number of genomic samples in which the allele is detected and the number of genomic samples analyzed.

12. The method of claim 1, wherein the RCG is prepared by performing degenerate oligonucleotide priming-polymerase chain reaction (DOP-PCR) using a degenerate oligonucleotide primer having a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes at least 7 TARGET nucleotide residues, wherein x is an integer from 0-9, and wherein each N is any nucleotide residue, and wherein the tag is a polynucleotide having from about 0 to about 20 nucleotides.

13. The method of claim 12, wherein the TARGET nucleotide sequence includes at least 8 nucleotide residues.

14. The method of claim 6, wherein the RCG is prepared by interspersed repeat sequence-polymerase chain reaction (IRS-PCR).

15. The method of claim 6, wherein the RCG is prepared by arbitrarily primed-polymerase chain reaction (AP-PCR).

16. The method of claim 6, wherein the RCG is prepared by adapter-polymerase chain reaction.

17. The method of claim 2, wherein at least a fraction of the SNP-ASO is labeled.

18. The method of claim 17, wherein an excess of a non-labeled SNP-ASO is added during the hybridization step, wherein the non-labeled oligonucleotide is complementary to a different allele of the same SNP than the labeled SNP-ASO.

19. The method of claim 17, further comprising performing a parallel hybridization reaction wherein the RCG is hybridized with a labeled SNP-ASO, wherein the oligonucleotide is complementary to a different allele of the same SNP than the labeled SNP-ASO.

20. The method of claim 19, wherein the two SNP-AGOs are distinguishably labeled.

21. The method of claim 17, an excess of non-labeled SNP-ASO is present during the hybridization.

22. The method of claim 2, wherein the SNP-ASO is composed of from about 10 to about 50 nucleotides residues.

23. The method of claim 22, wherein the SNP-ASO is composed of from about 10 to about 25 nucleotides residues.

24. The method of claim 17, wherein the label is a radioactive isotope.

25. The method of claim 24, further comprising the step of exposing the RCG to a film to produce a signal on the film which corresponds to the radioactively labeled hybridization products if the SNP is present in the RCG.

26. The method of claim 17, wherein the label is a fluorescent molecule.

27. The method of claim 26, further comprising the step of exposing the RCG to an automated fluorescence reader to generate an output signal which corresponds to the fluorescently labeled hybridization products if the SNP is present in the RCG.

28. The method of claim 17, wherein a plurality of SNP-ASOs are labeled with fluorescent molecules, each SNP-ASO being labeled with a spectrally distinct fluorescent molecule.

29. The method of claim 28, wherein the number of SNP-ASOs having a spectrally distinct fluorescent molecule is at least two.

30. The method of claim 28, wherein the number is selected from the group consisting of three, four and eight.

31. The method of claim 2, wherein a plurality of RCGs are labeled with fluorescent molecules, each RCG being labeled with a spectrally distinct fluorescent molecule, and wherein all of the RCGs having a spectrally distinct fluorescent molecule.

32. The method of claim 1, wherein the RCG is prepared by performing degenerate oligonucleotide priming-polymerase chain reaction using a degenerate oligonucleotide primer having a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes fewer than 7 TARGET nucleotide residues wherein x is an integer from 0 to 9, wherein each N is any nucleotide residues, and wherein the tag is a polynucleotide having from about 0-20 nucleotides.

33. The method of claim 32 wherein the TARGET nucleotide sequence includes at least 5 nucleotide residues.

34. The method of claim 32 wherein the TARGET nucleotide sequence includes at

least 6 nucleotide residues.

35. The method of claim 2, wherein the RCG is labeled.

36. The method of claim 4, wherein a plurality of different SNP-ASOs are attached to the surface.

37. The method of claim 1, wherein the RCG is prepared by performing multiple primed DOP-PCR.

38. The method of claim 2, wherein the genomic sample is characterized by generating a genomic pattern based on the presence or absence of the allele of the SNP in the genomic sample.

39. The method of claim 38, wherein the genomic pattern is a genomic classification code.

40. A method for characterizing a tumor, the method comprising:

isolating genomic DNA from tumor samples obtained from a plurality of subjects,

preparing a RCGs from each genomic DNA,

performing a hybridization reaction with a SNP-ASO and the plurality of RCGs, wherein the SNP-ASO is complementary to one allele of a SNP, and

characterizing the tumor based on whether the SNP-ASO hybridizes with at least some of the RCGs, whereby if the SNP oligonucleotide hybridizes with at least some of the RCGs, then the allele of the SNP is present in the genomic DNA of the tumor.

41. The method of claim 40, wherein the hybridization reaction is performed with a plurality of SNP-ASOs immobilized on a surface, and wherein the hybridization is performed on the plurality of RCGs, each RCG being analyzed separately.

42. The method of claim 40, wherein the RCGs are prepared by performing POP-PCR using a degenerate oligonucleotide primer having a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes at least 7 TARGET nucleotide residues and wherein x^1 is an integer from 0 to 9, wherein each N is any nucleotide residue, and wherein each tag is a polynucleotide having from 0 to about 20 nucleotide residues.

43. The method of claim 42, wherein the TARGET nucleotide sequence includes at least 8 nucleotide residues.

44. The method of claim 40, wherein the RCGs are PCR-generated RCGs.

45. The method of claim 40, wherein the RCGs are native RCGs.

46. The method of claim 40, wherein the RCG is prepared by performing DOP-PCR using a degenerate oligonucleotide primer having a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes has fewer than 7 TARGET nucleotide residues and wherein x is an integer from 0 to 9, wherein each N is any nucleotide residue, and wherein each tag is a polynucleotide having from 0 to about 20 nucleotide residues

47. A method for generating a genomic pattern for an individual genome, the method comprising:

preparing a RCG from the individual genome,

analyzing the RCG for the presence or absence of at least one SNP allele, and

generating a genomic pattern for the individual genome based on the presence or absence of SNP alleles.

48. The method of claim 47, wherein analyzing the RCG involves a hybridizing the RCG

with a panel of SNP-ASOs, each of which is complementary to one allele of a SNP, and identifying the genomic pattern by determining the ability of the RCG to hybridize with each SNP-ASO.

49. The method of claim 47, wherein the genomic pattern is a genomic classification code which is generated from the pattern of SNP alleles for each RCG.

50. The method of claim 49, wherein the genomic classification code is also generated using the allelic frequency of the SNPs.

51. The method of claim 47, wherein the genomic pattern is a visual pattern.

52. The method of claim 47, wherein the genomic pattern is a digital pattern.

53. The method of claim 48, wherein the SNP-ASOs are immobilized on a surface.

54. The method of claim 47, further comprising performing a parallel reaction wherein the hybridization reaction is performed using a panel of labeled complementary SNP-ASOs.

55. The method of claim 54, wherein the RCG is immobilized on a surface and wherein each SNP-ASO of the panel is hybridized with a separate surface.

56. The method of claim 54, wherein the RCGs is immobilized on a surface and wherein a plurality of SNP-ASOs of the panel are hybridized with a single surface, each SNP-ASO being labeled with a spectrally distinct fluorescent molecule.

57. The method of claim 47, wherein the RCGs is prepared by performing DOP-PCR using a degenerate oligonucleotide primer having a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes at least 7 TARGET nucleotide residues and

wherein x is an integer from 0 to 9, wherein each N is any nucleotide residue, and wherein each tag is a polynucleotide having from 0 to about 20 nucleotide residues.

58. The method of claim 47, wherein the RCG is a PCR-generated RCG.

59. The method of claim 47, wherein the RCG is a native RCG.

60. The method of claim 47, wherein the RCG is prepared by performing DOP-PCR using a degenerate oligonucleotide primer having a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes less than 7 TARGET nucleotide residues and wherein x is an integer from 0 to 9, wherein each N is any nucleotide residue, and wherein each tag is a polynucleotide having from 0 to about 20 nucleotide residues

61. A method for generating a genomic classification code for a genome, the method comprising:

preparing a RCG from the genome,

analyzing the RCG for the presence or absence of SNP alleles of known allelic frequency, and

identifying a genomic pattern of SNP alleles for the RCG by determining the presence or absence therein of SNP alleles, and

generating a genomic classification code for the RCG based on the presence or absence and the allelic frequency of the SNP alleles.

62. The method of claim 61, wherein the RCG is hybridized reaction with a panel of SNP-ASOs of known allelic frequency, each of which is complementary to one allele of a SNP, and identifying the genomic pattern based on whether each SNP-ASO hybridizes with the RCG.

63. The method of claim 62, wherein the SNP-ASOs are immobilized on a surface.

64. The method of claim 62, wherein the RCG is immobilized on a surface.

65. The method of claim 61, wherein the RCG is prepared by performing POP-PCR using a degenerate oligonucleotide primer having a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes at least 7 TARGET nucleotide residues and wherein x is an integer from 0 to 9, wherein each N is any nucleotide residue, and wherein each tag is a polynucleotide having from 0 to about 20 nucleotide residues.

66. The method of claim 61, wherein the RCG is a PCR-generated RCG.

67. The method of claim 61, wherein the RCG is a native RCG.

68. The method of claim 61, wherein the RCG is prepared by performing DOP-PCR using a degenerate oligonucleotide primer having a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes less than 7 TARGET nucleotide residues and wherein x is an integer from 0 to 9, wherein each N is any nucleotide residue, and wherein each tag is a polynucleotide having from 0 to about 20 nucleotide residues.

69. A composition, comprising:
a plurality of RCGs immobilized in an ordered array on a surface.

70. The composition of claim 69, wherein the RCGs prepared by the method of claim 125.

71. The composition of claim 69, wherein the RCGs are PCR-generated RCGs.

72. The composition of claim 69, wherein the RCGs are native RCGs.

73. A kit, comprising:

100. The composition of any one of 90-95, wherein x is 9.

101. A method for identifying a SNP, the method comprising:

preparing a set of primers from a RCG, wherein the RCG comprises a set of polymerase chain reaction (PCR) products,

performing PCR using the set of primers on at least one of isolated genome to produce a set of DNA products, and

identifying a SNP on the set of DNA products.

102. The method of claim 101, wherein the plurality of isolated genomes is a pool of genomes.

103. The method of claim 101, wherein the isolated genomes are RCGs.

104. The method of claim 103, wherein the RCG is prepared by DOP-PCR.

105. The method of claim 101, wherein the step of preparing the set of primers is performed by at least the following steps:

preparing a RCG and separating the set of PCR products in the RCG into individual PCR products,

determining the sequence of each end of at least one of the PCR products, and

generating primers for use in the subsequent PCR step based on the sequence of the ends of the inserts.

106. The method of claim 105, wherein the set of PCR products are separated by gel electrophoresis.

107. The method of claim 106, further comprising the step of preparing libraries from

segments of the gel containing several PCR products and isolating clones from the library, each clone including a PCR product containing plasmid from the library.

108. The method of claim 105, wherein the set of PCR products are separated by high pressure liquid chromatography.

109. The method of claim 105, wherein the set of PCR products are separated by column chromatography.

110. The method of claim 101, wherein the RCG is prepared by performing DOP-PCR using a degenerate oligonucleotide primer having a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes at least 7 TARGET nucleotide residues and wherein x is an integer from 0 to 9, wherein each N is any nucleotide residue, and wherein each tag is a polynucleotide having from 0 to about 20 nucleotide residues.

111. The method of claim 110, wherein the TARGET nucleotide sequence includes 8 nucleotide residues.

112. The method of claim 110, wherein the TARGET nucleotide sequence includes 9 nucleotide residues.

113. The method of claim 110, wherein the TARGET nucleotide sequence includes 10 nucleotide residues.

114. The method of claim 110, wherein the TARGET nucleotide sequence includes 11 nucleotide residues.

115. The method of claim 110, wherein the TARGET nucleotide sequence includes 12

nucleotide residues.

116. The method of claim 101, wherein the RCG is prepared by IRS-PCR..

117. The method of claim 101, wherein the RCG is prepared by AP-PCR.

118. The method of claim 101, wherein the RCG is prepared by adapter-polymerase chain reaction.

119. The method of claim 101, wherein the RCG is prepared by performing DOP-PCR using a degenerate oligonucleotide primer having a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes less than 7 TARGET nucleotide residues and wherein x^1 is an integer from 0 to 9, wherein each N is any nucleotide residue, and wherein each tag is a polynucleotide having from 0 to about 20 nucleotide residues.

120. The method of claim 101, wherein x is greater than one.

121. The method of claim 101, wherein the first and second steps of PCR products are generated using the same primers.

122. A composition comprising:

a panel of SNP-ASOs immobilized on a surface, wherein the SNP-ASOs are prepared by the method of claim 101.

123. The composition of claim 122, wherein each SNP-ASO is immobilized in a discrete area of the surface.

124. The composition of claim 122, further comprising a panel of complementary SNP-ASOs immobilized on discrete areas of the surface.

125. A method for obtaining a RCG using DOP-PCR, the method comprising:

performing DOP-PCR using a degenerate oligonucleotide primer having a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes at least 7 TARGET nucleotide residues and wherein x is an integer from 0 to 9, wherein each N is any nucleotide residue, and wherein each tag is a polynucleotide having from 0 to about 20 nucleotide residues.

126. The method of claim 125, wherein the TARGET nucleotide sequence includes 8 nucleotide residues.

127. The method of claim 125, wherein the TARGET nucleotide sequence includes 9 nucleotide residues.

128. The method of claim 125, wherein the TARGET nucleotide sequence includes 10 nucleotide residues.

129. The method of claim 125, wherein the TARGET nucleotide sequence includes 11 nucleotide residues.

130. The method of claim 125, wherein the TARGET nucleotide sequence includes 12 nucleotide residues.

131. The method of any one of 125-130, wherein x is from 3 to 9.

132. The method of any one of 125-130, wherein x is 6.

133. The method of any one of 125-130, wherein x is 7.

134. The method of any one of 125-130, wherein x is 8.

135. The method of any one of 125-130, wherein x is 9.

136. The method of claim 125, wherein the tag includes 6 nucleotide residues.

137. The method of any one of 125-136, further comprising using the RCG in a genotyping procedure.

138. The method of any one of 125-136, further comprising analyzing the RCG to detect a polymorphism.

139. The method of claim 138 wherein the RCG is analyzed using mass spectroscopy.

140. A method for assessing whether a subject is at risk for developing a disease, the method comprising:

preparing a RCG from a genomic sample obtained from the subject and characterizing the sample by the method of claim 1, whether one sample based on the presence or absence in the sample of a plurality of SNP alleles that occur in at least 10% of genomes obtained from individuals afflicted with the disease occur in the reduced subject complexity genome.

141. A method for identifying a set of SNP alleles associated with a disease, the method comprising:

preparing individual RCGs obtained from subjects afflicted with a disease using the same set of primers to prepare each RCG, and

a container housing a set of polymerase chain reaction primers for reducing the complexity of a genome, and

a container housing a set of SNP-ASOs, wherein the SNPs are present with a frequency of at least 50% in a RCG made using the set of primers.

74. The kit of claim 73, wherein the SNP-ASOs are attached to a surface.

75. The kit of any one of claims 73 or 74, wherein the set of polymerase chain reaction primers are primers for DOP-PCR.

76. The kit of claim 75, wherein the degenerate oligonucleotide primer has a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes at least 7 TARGET nucleotide residues and wherein x is an integer from 0 to 9, wherein each N is any nucleotide residue, and wherein each tag is a polynucleotide having from 0 to about 20 nucleotide residues.

77. The kit of claim 76, wherein the TARGET nucleotide sequence includes at least 8 nucleotide residues.

78. The kit of claim 76, wherein the TARGET nucleotide sequence includes at least 9 nucleotide residues.

79. The kit of claim 76, wherein the TARGET nucleotide sequence includes at least 10 nucleotide residues.

80. The kit of claim 76, wherein the TARGET nucleotide sequence includes at least 11 nucleotide residues.

81. The kit of claim 76, wherein the TARGET nucleotide sequence includes 12

nucleotide residues.

82. The kit of any one of claims 73 or 74, wherein the set of polymerase chain reaction primers are primers for ISR-PCR.

83. The kit of any one of claims 73 or 74, wherein the set of polymerase chain reaction primers are primers for AP-PCR.

84. The kit of any one of claims 73 or 74, wherein the set of polymerase chain reaction primers are primers for adapter-polymerase chain reaction.

85. The kit of any one of claims 73 or 74, wherein the SNP-ASOs are composed from 10 and 50 nucleotide residues.

86. The kit of any one of claims 73 or 74, wherein the SNP-ASOs are composed of from 10 and 25 nucleotide residues.

87. The kit of any one of claims 73 or 74, wherein the SNP-ASOs are labeled with a fluorescent molecule.

88. The kit of claim 75, wherein the degenerate oligonucleotide primer has a tag-$(N)_x$-TARGET nucleotide sequence, wherein the TARGET nucleotide sequence includes fewer than 7 TARGET nucleotide residues and wherein x is an integer from 0 to 9, wherein each N is any nucleotide residue, and wherein each tag is a polynucleotide having from 0 to about 20 nucleotide residues.

89. The kit of claim 73, wherein the set of polymerase chain reaction primers are primers for multiple-primed DOP-PCR.

90. A composition comprising:

a plurality of RCGs immobilized on a surface, wherein the RCGs are composed of a plurality of DNA fragments, each DNA fragment comprising a $(N)_x$-TARGET nucleotide portion, wherein the nucleotide sequence of TARGET is identical in each of the DNA fragments, wherein TARGET is a polynucleotide consisting of at least 7 nucleotide residues, wherein x is an integer from 0 to 9, and wherein N is any nucleotide residue.

91. The composition of claim 90, wherein the TARGET nucleotide sequence includes 8 nucleotide residues.

92. The composition of claim 90, wherein the TARGET nucleotide sequence includes 9 nucleotide residues.

93. The composition of claim 90, wherein the TARGET nucleotide sequence includes 10 nucleotide residues.

94. The composition of claim 90, wherein the TARGET nucleotide sequence includes 11 nucleotide residues.

95. The composition of claim 90, wherein the TARGET nucleotide sequence includes 12 nucleotide residues.

96. The composition of any one of claims 90-95, wherein x is from 3 to 9.

97. The composition of any one of 90-95, wherein x is 6.

98. The composition of any one of 90-95, wherein x is 7.

99. The composition of any one of 90-95, wherein x is 8.

comparing individual genetic loci in the RCGs with the same individual genetic loci in normal subjects to identify SNP associated with the disease.

142. A digital information product for representing genomic information, the product comprising:

a computer-readable medium having computer-readable signals stored thereon, wherein the signals define a data structure, the data structure including one or more data components, wherein each data component includes:

a first data element defining a genomic classification code that identifies a corresponding genome, and wherein each genomic classification code classifies the corresponding genome based one or more single nucleotide polymorphisms of the corresponding genome.

143. The difital information proiduc of claim 142, wherein the genomic classification code is a unique identifier of the corresponding genome.

144. The digital information product of claim 142, wherein the genomic classification code is based on a pattern of the single nucleotide polymorphisms of the corresponding genome, the pattern indicating the presence or absence of each single nucleotide polymorphism.

145. The digital information product of claim 142, wherein each data component also includes:

one or more data elements, each data element defining an attribute of the corresponding genome.

146. A process for making a digital information product comprising computer data signals defining a genomic classification code for a genome, the process comprising:

preparing a reduced complexity genome,

performing a hybridization reaction with the reduced complexity genome and at least one surface having a panel of single nucleotide polymorphism oligonucleotides immobilized thereon,

identifying a genomic pattern of single nucleotide polymorphisms for the reduced complexity genome by determining the presence therein of single nucleotide polymorphisms based on whether each single nucleotide polymorphism oligonucleotide hybridizes to the reduced complexity genome,

generating a genomic classification code for the reduced complexity genome based on the genomic pattern of the single nucleotide polymorphisms, and

encoding the genomic classification code as one or more computer data signals on a computer-readable medium.

147. A process for making a digital information product comprising computer data signals defining a genomic classification code for a genome, the process comprising:

preparing a reduced complexity genome,

performing a hybridization reaction with a panel of single nucleotide polymorphism oligonucleotides of known allelic frequency and a surface having the reduced complexity genome immobilized thereon,

identifying a genomic pattern of single nucleotide polymorphisms for the reduced complexity genome by determining the presence therein of single nucleotide polymorphisms based on whether each single nucleotide polymorphism oligonucleotide hybridizes to the reduced complexity genome,

generating a genomic classification code for the reduced complexity genome based on the pattern and the allelic frequency of the single nucleotide polymorphisms, and

encoding the genomic classification code as one or more computer data signals on a computer-readable medium.

148. A method for performing linkage analysis, comprising:

preparing individual RCGs obtained from members of one or more families ,

determining the presence or absence of SNP alleles in the RCGs, and

comparing the RCGs of the family members by comparing the presence or absence of the SNP alleles in the RCGs of the family members.

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